

## **GENETICS AND NEWBORN SCREENING ADVISORY COUNCIL MEETING**

The Genetics and Newborn Screening Advisory Council meeting was held on Friday, July 25th, 2014 at the Florida Department of Health Bureau of Laboratories, 1217 Pearl St., Jacksonville, FL.

### **Call to Order:**

The meeting was called to order at 10:04am EST by Paul Pitel, MD, Chairman of the Council. Roll was taken and introductions were made.

### **Members Present:**

Paul Pitel, MD, Chairman, Jacksonville  
Roberto Zori, MD, University of Florida  
Susanne Crowe, MHA, Jacksonville  
Robert Fifer, PhD, Miami (UM)  
Elena Perez, MD, Miami (UM)  
Heather Smith, Lakeland  
David Auerbach, MD, Orlando  
Dorothy Shulman, MD, St. Petersburg (USF)  
Bonnie Hudak, MD, Jacksonville  
Melissa Perez, Tallahassee (via teleconference)  
Lori Kohler, RN, representing Lori Gephart, RN, Tallahassee (via teleconference)

### **Guests:**

Jeanne Brunger, PerkinElmer, Inc.  
Shana Bauer, Audiology Extern, University of Miami  
Chelsey DeFour, Audiology Extern, University of Miami  
Cynthia Robinson, Clarke Schools for Hearing and Speech, Jacksonville  
Larry Vroegindewey, DOH SCID  
Lucy Raub, University of Florida  
Sharon Bowden, Pediatrix Medical Group  
Yorquiz PerezMendez, Pediatrix Medical Group  
George Fox  
Barry Byrne, MD  
Dana Ferrell  
Gerald Schiebler  
Helen Burgess, University of Florida

### **DOH Personnel Present:**

Lois Taylor, RN, CMS, Tallahassee  
Whitney G. Jones, CMS, Tallahassee  
Drew Richardson, CMS, Tallahassee  
Pam Tempson, CMS, Tallahassee  
Linda Deterding, RN, CMS, Tallahassee  
Ming Chan, PhD, Bureau of Laboratories, Jacksonville

## **Housekeeping/Reminders**

Dr. Pitel reminded council members and guests to hand in their money for lunch and to review the minutes of the February 2014 council meeting for approval after the break.

## **2014 Legislative Update**

Ms. Taylor gave the legislative update. Some bills that passed through Children's Medical Services (CMS) are HB 561 that dealt with attorneys for dependent children with special needs, HB 989 dealt with human trafficking, HB 7141 also referred to human trafficking, HB 977 referred to motor vehicle insurance and driver education for children in care, and SB 1666 referred to child welfare and was a significant bill for CMS.

The Department submitted legislation to correct technical errors or make technical changes in the Newborn Screening (NBS) statute 383.14. In 2004 language was added to the statute so CMS could provide newborn screening reports to primary care physicians (PCP), but this limited who could receive these reports to only the PCP and their employees. Changes were submitted to amend the language to say "health care practitioner which includes many other health care professionals. The change was passed and now these practitioners can now enroll in the Florida Newborn Screening Results website (FNSR) and get screening results as of July 1, 2014.

Small technical changes needed to be made to the Newborn Screening Hearing statute 383.15 such as public law records and removal of the implementation date of hearing screening in Florida. As this bill was going through the process in the legislature a portion was added, 383.16, which requires audiologists to ask parents of a child newly diagnosed with hearing loss if they would like receive information from Early Steps providers. The Department of Health has compiled a list of providers that audiologists can use to give to parents if they would like information from these providers. This has already been implemented and the Department has one more step to complete which is to update the CMS data system so it's automatic. Dr. Fifer expressed that parents had become frustrated with the lack of information on providers across the state as they were only receiving information on providers within their immediate area and this change solves that frustration. Audiologists also expressed concern over the amount of work this change would entail, but after discussion it was concluded that it would only require some work up front to prepare for the implantation.

Other bills that were introduced that affected Newborn Screening are HB 1163 which would add adrenoleukodystrophy (ALD) screening to the screening to the panel. This did not pass and there was no companion bill but the Department will reach out to Representative La Rosa to continue the discussion about ALD. Two memorial bills were also introduced, CS/HM 1165 and SB 1288, which asked the Florida Legislature to send the memorial bills to the US Congress asking to add ALD to the National Recommended Uniform Screening Panel. Both of these bills did not pass. Dr. Pitel expressed concern that the process of implementation of screening for new disorders was being bypassed.

## **SCID Update**

Ms. Taylor gave the update for SCID. There was a suggestion made by the Council members to send a Thank You letter to Representative Matt Hudson. Rep. Hudson was actively involved in adding SCID to the newborn screening panel through supporting the request for budget authority through the Legislature. Dr. Pitel suggested a motion to complete and send the letter to Rep. Hudson. Dr. Fifer approved the motion and it was seconded. The council's vote was unanimous.

The number of presumptive positive results for SCID in 2013 is 29. The latest numbers so far for 2014 is 8. There are still a high number of inconclusive results due to premature babies in the NICU. The program has added some categories to the case reports to distinguish between the four variants of x-linked SCID. Other states were found to be counting Chylothorax cases. Ms. Taylor asked the council if they would like to continue counting and keeping data for Chylothorax cases found through this screening. Dr. Perez and Dr. Auerbach agreed with keeping track of the Chylothorax cases found as it indicates what is being found through the SCID screening and may be useful in the future. Dr. Perez spoke to evolution of the process and flow chart since SCID screening was implemented. If there are any cases that have prior normal screenings then the case is closed as normal. These cases were creating some unnecessary referrals and since that has changed there have been less of them. It is also important to ask what has been going on with mom that may have affected babies and show abnormal screenings.

Dr. Perez asked the council about the impact of mothers receiving a devastating diagnosis for their baby and the impact it has on post-partum depression and if the council could possibly address this issue. Dr. Pitel stated that providing support for these families takes time and effort and is not billable. It was suggested that support groups are beneficial in this situation.

## **CCHD Implementation Update**

Ms. Taylor gave the update on CCHD implementation. A summary sheet was developed by the CCHD workgroup and was distributed to all providers in the state. 100% of the 119 hospitals in the state are doing the CCHD screening. Close to 75% of birthing centers are performing the screening for having a physician do the screening at the well-baby checkup. Data has only started to be collected and of the six (6) cases, three (3) turned out to be normal, one had an error in reporting, one was diagnosed as a cardiac other where the screen showed up as failed, and one was screened and diagnosed with CCHD. A Midwife in the state has volunteered to become a cheerleader for CCHD after she experienced delivering a well-baby but two days later the baby showed signs of CCHD, was screened, then flown to a cardiac center and diagnosed with CCHD. The Midwife has agreed to help encourage other birthing centers in implementing the CCHD screening.

Dr. Schiebler introduced Dr. Barry Byrne as the person who first voiced the need for CCHD screening in Florida. Dr. Schiebler made a suggestion to change the current referral process so that any babies screened with possible CCHD should be referred to a CMS cardiac center. Dr. Schiebler also proposed for a study, led by Dr. Mark Hudak, to be done on one site verses

two site CCHD screening. Dr. Pitel indicated that an IRB should be proposed and sent to the Florida Department of Health for review and that the advisory council is open to a study pending IRB approval. Dr. Pitel moved to have the cardiac workgroup together for a conference call to discuss the current referral process. Dr. Shulman seconded and the vote was unanimous.

### **Newborn Screening Laboratory Update**

Ms. Crowe gave the Newborn Screening Laboratory Update to the council. The current vacancies at the lab are an NBS Lab Director, Chem. Administrator II, and a Chemist III. The lab is currently working on recruiting. Technical staff has reduced, decreasing from 28 to 21 staff. The lab recently hired a Quality Assurance office for the lab. The lab had a CLIA inspection last week and the verbal report indicated that there were two minor deficiencies but otherwise went well. There has been some staff turnover and workers being out on workers comp which caused delays in reporting. The Newborn Screening staff traveled from Tallahassee to Jacksonville to help catch the lab up. A Biochemical geneticist was hired to help the lab ensure that testing is done correctly.

Dr. Pitel spoke about University of Miami (UM) lab issues. The UM laboratory is no longer available for confirmatory testing for the state and Dr. Bodamer is no longer running the lab. Ms. Taylor stated that some of the genetic centers were submitting some samples to the UM lab for testing but not many. The Department may pursue other providers if there are any available for this kind of testing.

Minutes from the February 2014 meeting were approved by the council.

### **Speaker: Pompe Update**

Dr. Byrne gave an update on Pompe Disease. Dr. Byrne presented data from Taiwan who screens for Pompe disease. About 500,000 babies have been screened for Pompe in Taiwan by the time this data was published and suggests that early treatment of the disease has better outcome results. Dr. Byrne suggested that there is the potential, with sponsorship, to do a retrospective analysis of existing blood specimens. Pompe Disease is not yet added to the Recommended Uniform Screening Panel. Secretary Siebelius ordered for an independent report to be due at the end of July. Dr. Pitel asked what the status of blood specimens are in Florida. Ms. Taylor stated that blood specimens were destroyed after six months. IRB approval would have to be obtained to be able to use de-identified blood specimens for a study or other testing. The group discussed the possible costs of treatment and ethical problems associated with adding Pompe to the screening panel. Dr. Pitel made a motion to later discuss submitting an IRB for a study of Pompe. Dr. Zori seconded and the motion was approved unanimously.

### **Invited Speaker: Future of Newborn Screening Specialty Services**

Dana Ferrell led the discussion regarding the additional funding that was provided to the three Genetic Referral Centers for children diagnosed with genetic conditions. The Legislature provided non-recurring funds of \$600,000 to the three genetic centers for one year. There was a following discussion regarding the future need for more resources for genetic services. Dr. Pitel made a motion to support the need for future funding of the genetic centers. Dr. Shulman seconded and the motion was approved unanimously.

### **Newborn Screening Follow-Up Program Update**

Pam Tempson gave a Hearing Follow-up update to the council. Ms. Tempson asked the council to create and utilize a workgroup to meet grant expectations for the hearing program. One of the hearing program grants requires the creation of a stakeholder group which is the basis of this request. Dr. Pitel approved the motion and Dr. Shulman and Dr. Zori seconded. The motion was approved unanimously.

Ms. Tempson stated that eReports was showing great improvement in usage at 80% in reporting through eReports. There are only 14 hospitals left to train in eReports in the state. Once this is done every hospital will have at least one person trained in using eReports. 2013 hearing data showed that 96.44% of babies in Florida were screened for hearing loss. 2013 Pass vs. Not Pass data shows that 199,950 babies passed their initial screening and 8,023 did not pass the initial screening. Of those 8,023, 6,537 passed a subsequent test, 1,085 are still pending, 187 were diagnosed with permanent hearing loss, and the rest were closed to other reasons.

Drew Richardson gave an update on Newborn Screening Systems to the council. Before Florida Newborn Screening Results (FNSR) went live in 2009 the Newborn Screening Follow-up Program was processing over 40,000 physician requests per year. Now that FNSR is in place the follow-up program is processing below 5,000 requests per year and the website is processing over 120,000 requests per year. Also, this year there was a change in the statute language that now allows other health care providers to use this website rather than only primary care providers.

Electronic Laboratory Ordering/Electronic Laboratory Reporting (ELO/ELR) is continuing to be a work in progress. The NBS program is working with about ten hospitals on file layout and testing data transfers. The ELO/ELR project is currently on hold while the NBS program is undergoing a laboratory information system upgrade to Screening Center. Screening Center was set to go-live in June of 2014, but was pushed back due to flaws in the program. Future date for Screening Center go-live is tentatively set for some time in August.

Direct Secure Messaging (DSM) is a system that the NBS follow-up program used to route protected health information to providers. However, DSM usage was terminated in mid-July. The follow-up program is now using Move-IT as the replacement for DSM. Critical Congenital Heart Disease (CCHD) implementation began just a few weeks ago.

CCHD results have begun being entered into the system on 7/1/2014 and by the next council meeting there should be some data to report.

Lois Taylor informed the council that a new CLIA regulation where laboratories can release results directly to patients. The NBS Program must have a policy procedure in place for this regulation by October 6. Lois also informed the council regarding some challenges with Medicaid Managed Care (MMA) and receiving reimbursement from Medicaid billing. Agency for Healthcare Administration (AHCA) is looking into letting the program bill them directly instead of Medicaid HMO's.

### **New Discussion Items**

Bonnie Hudak informed the council that the lab received a grant from Nemours Jacksonville for NBS video educational material for Cystic Fibrosis.

### **Public Comments/closing/summary**

Dr. Gerald Schiebler gave a public comment regarding Medicaid Managed Care (MMA) and funding issues. Dr. Pitel thanked Dr. Schiebler for his comment.

### **Adjournment**

The meeting adjourned at 2:20pm EST.